Health Professional Information for Cornelia de Lange Syndrome (CdLS) [Brachman-de Lange Syndrome]

CdLS is a rare, usually sporadic, dysmorphic syndrome with a birth incidence of between 1 in 40,000 and 1 in 100,000. The diagnosis is clinical and usually based on the combination of distinctive facial appearance, limb anomalies and prenatal onset short stature. Misdiagnoses are not uncommon and this care card assumes that a firm diagnosis of CdLS has been made by an experienced dysmorphologist. The aim is to highlight treatable complications of the disorder and to provide a record for health professionals of all clinicians involved with each patient.

Growth and Endocrine Problems:
Almost all children with CdLS show both pre and postnatal **growth failure** in spite of adequate diet. The use of specific **CdLS growth charts** is strongly recommended and these are available at no cost on-line at **http://www.cdls.org.uk/background**. Therapy with recombinant growth hormone is generally considered non-beneficial. **Cryptorchidism** and **micropenis** are relatively common in males. Puberty can be slightly delayed in both sexes but may be incomplete. **Primary amenorrhoea** is common.

Gastrointestinal Problems:
Gastro-oesophageal reflux disease (GORD) is very common and under-recognised. Undiagnosed GORD may be an explanation for behavioural abnormalities, pain, self abuse recurrent respiratory disease or other puzzling symptoms and signs. Many children benefit from endoscopy and/or a therapeutic trial of medication such as a proton pump inhibitor. **Fundoplication** may be required. **Barrett's oesophagus** and **Sandifer complex** can occur. A low threshold for referral to a paediatric GI specialist is recommended. Acute surgical presentation may be due to **caecal volvulus** and **malrotation** with nonfixation of the caecum and ascending colon. **Diagphragmatic hernia**, **duodenal atresia** and **annular pancreas** are also non-randomly associated with CdLS.

Neurological, Learning and Behavioral Problems:
Learning disability of variable severity is almost universal. Skills charts are available at no cost on-line at **http://www.cdls.org.uk/background**. **Expressive language delay** is particularly common and early use of signing is recommended. **Hypertonicity** and **hyperreflexia** are common. There may be signs of an **autonomic neuropathy**. Some children have specific behavioural problems including **autistic-like features** and **self injury**.

Unusual **hand posturing** at eye level is very common. **Seizures** may be under-recognised and non-convulsive seizure activity has been observed.

Orthopaedic Problems:
Bilateral upper and lower **distal limb malformations** are very common but highly variable, ranging from **small hands** with proximally placed thumbs to **oligodactyly** with severe hypoplasia of the forearms. **Fixed flexion contractures** of the elbow are common. Even with severe anomalies the manipulative abilities are often remarkably good. **Hip dysplasia** may be a significant and under-recognised problem in CdLS.

Oro-dental Problems:
Cleft palate occurs in ~20% of cases although **submucous clefting** is probably under-recognised. Nasopharyngeal airways tend to be narrow. Micrognathia, microstomia and **dental crowding** are very commonly associated. Dental caries may be problematic and periodic examination and treatment under anaesthesia is often required.

Ophthalmology and Audiology:
Ptosis, recurrent blepharoconjunctivits and **myopia** are common in CdLS. Ptosis surgery may assist in motor development. Baby shampoo eyelash scrubs may be helpful in relieving recurrent red eye discharge although some children may require nasolacrimal duct probing for obstruction. **Nystagmus**, **mild microcornea**, and/or **strabismus** can occur. **Cataract** and **glaucoma** have been rarely reported and may represent chance occurences or the result of self-injury. Children with severe myopia may be at higher risk for **retinal detachment**. Optic **nerve pallor** is uncommon although a visually insignificant **pigmented ring around the optic nerve** is almost universal. Hearing loss is very common and assessment by a paediatric audiologist is recommended. External auditory canal stenosis is very common.

Cardiac Problems:
Congenital heart disease has been reported in 20% of cases, usually **ventricular septal defects** or **pulmonary artery stenosis**. **Perioral blueness** is very common and may not, in itself, require further investigation.

Dermatology:
Cutis marmorata and **generalised hirsuitism** are very common features of CdLS and do not merit further investigation.

Updated December 2002, Dr David FitzPatrick, MRC Senior Clinician Scientist and Honorary Consultant in Clinical Genetics, Western General Hospital, Edinburgh. Medical Director CdLS UK & Ireland. With contibutions from Dr. Toni Kline (Genetics, Baltimore), Dr Doug Clements (Dental, Baltimore), Dr. Alex V. Levin (Ophthalmology, University of Toronto), Dr Peter Gillett (GI, Edinburgh) and Prof. Chris Oliver (Psychology, Birmingham)

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Further information

Further information on CdLS can be obtained from
the CdLS Foundation UK and Ireland
Tall Trees
106 Lodge Lane
Grays
Essex RM16 2UL

Tel/Fax: 01375 376439

e-mail: info@cdls.org.uk

Web: www.cdls.org.uk

The Foundation is the support group for carers and professionals involved in the life of the person with CdLS. The organisation is part of an international federation of CdLS groups and participates in the international Scientific Advisory Council (SAC) which features specialists from around the world with particular emphasis

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Personal Details

Name _____

DoB _____

Address _____

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Phone _____

Emergency Contact

Name _____

DoB _____

Address _____

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Phone _____

MEDICAL ALERT

The bearer of this card has **CORNELIA DE LANGE SYNDROME** a condition that may cause the following:

A. Growth and Endocrine Problems
B. Gastro-intestinal problems
C. Seizures
D. Cardiac problems
E. Neurological, learning and behaviourial problems
F. Oro-dental problems

CORNELIA DE LANGE SYNDROME

CdLs care card

GP	Prescription	
Name	Date	Date
Surgery		
Phone		
Date		

Optician	Dentist
Name	Name
Address	Address
Phone	Phone
Date	Date

Cardiologist	Gastrologist
Name	Name
Address	Address
Phone	Phone
Date	Date

Geneticist	Neurologist
Name	Name
Hospital	Hospital
Phone	Phone
Date	Date

Specialist	Specialist
Name	Name
Address	Address
Phone	Phone
Date	Date

Specialist	Specialist
Name	Name
Address	Address
Phone	Phone
Date	Date

Using your Cdls Care Card

The CdLS care card has been developed to enable the multi-disciplinary team necessary for the best management of the syndrome to be aware of the actions of other specialists.
Carers should complete the details every time the patient sees a specialist. By listing the contact details and the date of visit, other specialists can get in touch.

By listing prescriptions, your doctors can be confident that there are no conflicting treatments being recommended.
Other specialists may include: psychologist, occupational therapist, anaesthetist, ophthalmologist, paediatrician, social worker, surgeon and speech therapist. Carers should make sure they indicate the speciality of the professional.

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